Genomics Education Programme

NHS
Health Education England

Identifying the clinical clues of genetic conditions

Some clinical features make it more likely that a condition has a genetic cause. What clues do you need to consider?

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There are clinical clues which could suggest that a family has an inherited susceptibility to a common disease, or that a condition has a strong genetic component. Some things to consider are:

- Multiple closely related individuals with the same condition, particularly if the condition is rare
- Disorders which occur at an earlier age than usual (especially if onset is early in multiple family members.) Examples include:
    - Breast cancer <45-50 years (premenopausal)
    - Colon cancer <45-50 years
    - Prostate cancer <50-60 years
    - Vision loss <50 – 60 years
    - Dementia <60 years
    - Cataracts
    - Deafness (congenital or juvenile)
    - Blindness (congenital or juvenile)
- Sudden cardiac deaths in people who seemed healthy
- Individual or couple with three or more pregnancy losses (e.g. miscarriages, stillbirths)
- Medical problems in the offspring of parents who are related by blood
- Multiple congenital anomalies or dysmorphic features, particularly if associated with developmental delay.

There is a network of specialist genetic services available to help provide support and advice – please contact the Genomics Education Programme for more information about your local centre.

For further information on genetic conditions, the GEP has produced a series of conditions factsheets which are available to download from our website:

www.genomicseducation.hee.nhs.uk